Case Report
JCAM

Schwartz-jampel syndrome; case report

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Abstract

Schwartz-Jampel Syndrome is a rare disease that is characterized with skeletal deformities, joint contractures, and dysmorphic facial appearance. Most patients become symptomatic within the first decades of life. The diagnosis is based on clinical findings. Movement limitation of joints, delay in starting to walk, and walking with support are common preliminary findings. The disease is caused by the mutation in heparan sulfate proteoglycan 2 (HSPC2) gene which encodes perlecan protein. Herein we report a case of Schwartz-Jampel syndrome with the age of 18.

Keywords

Schwartz-Jampel Syndrome; Skeletal Deformity; Heparan Sulfate Proteoglycan 2; Perlecan Protein

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Introduction

Schwartz-Jampel Syndrome (SJS) is a rare disease that characterized with skeletal deformities, joint contractures, and dysmorphic facial appearance. Although, we have very few diagnosed cases with SJS in the literature, there has been a certain accumulation of knowledge about the disease since its first definition in 1962. As the genetic and phenotypic characteristics of the disease are well known, the intrauterine diagnosis will become common and new treatment options will be developed accordingly. This article has been written to draw attention to the rare disease and contribute to the literature.

Case Report

An 18-year-old male admitted to the outpatient clinic with the complaint of difficulty in walking. Prenatal and natal developmental stages were normal, and it was noted that limitation on arms and legs started when the patient was at the age of one. He started to walk lately and could walk with support. He had been operated due to joint deformities and became unable to walk in time. His parents are first-degree relatives, and his brother died at the age of 4.5 because of the same illness. Physical and neurological exam; he was 5 feet tall, dysmorphic facial appearance, bilateral low-set ear, microphthalmia, micrognathia, distorted tooth shape, scoliosis towards the right in the lumbar and thoracic spine areas, genu valgus, quadriparesis (MRC grade 3 on upper limbs, MRC grade 2 on lower limbs), absence of deep tendon reflexes, sensory examination was normal, and he was mobile with a wheelchair (Figure 1).

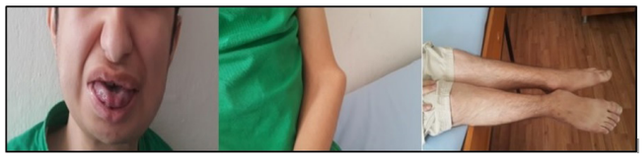

Figure 1. Dysmorphic facial appearance and joint deformities

Complete blood count, biochemical panel, hormone profile, and creatine phosphokinase (CPK) were all normal. Infectious and vasculitic tests were normal. Levels of lactic and pyruvic acid in the blood and amino acids in urine were normal. Radiographs revealed that scoliosis towards to the right in the lumbar and thoracic spine areas, common skeletal deformities, and joint contractures. (Figure 2)

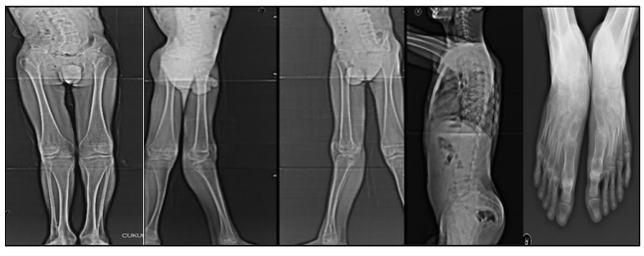

Figure 2. Common skeletal deformities and joint contractures shown on radiography

There were no significant pathologic findings on Electrocardiography and echocardiography. Electroneuromyography (ENMG) revealed normal nerve conduction velocities, persistent electrical activity, and myotonic discharges.

Discussion

SJC, also known as chondrodystrophic myotonia, was first described by Schwartz and Jampel in 1962 in two children with a typical phenotypic appearance of the disease [1]. It is estimated that the prevalence of the disease is less than 1 per 1.000.000. Less than 100 cases have been reported up to now [2]. Most patients become symptomatic within the first decades of life. The diagnosis is based on clinical findings. Movement limitation of joints, delay in starting to walk, and walking with support are common preliminary findings [2]. As muscle spasms and joint deformities increase, late-onset walking gradually deteriorates and most patients become unable to walk before the age of ten [2,3]. Short length, mask face, ophthalmoparesis, micrognathia, myopia, flat face, low-set ears, small jaw, deterioration of tooth structure, short neck, kyphosis, scoliosis, movement limitation of joints, continuous muscle activity, hypertrophic muscles and muscle spasms are the common clinical findings [1-3]. Also, hydrocephalus, carpal tunnel syndrome, myelopathy and occasionally mental retardation may be seen [3]. The SJS gene is on chromosome 1 (1p34-p36) [4,5]. Although the disease often shows autosomal recessive inheritance, there are cases of autosomal dominant inheritance [5]. The disease is caused by the mutation in heparan sulfate proteoglycan 2 (HSPC2) gene which encodes perlecan protein [6]. Perlecan protein is a proteoglycan in the cartilage and muscle membrane. Loss of function causes abnormal ion channel expression and aggregation of acetylcholinesterase [6]. Intrauterine diagnosis is possible. Ultrasonographic examination performed in the second trimester reveals that the fingers are constantly in a state of flexion, lack of movement, short and springy femur [3-6]. Electromyography (EMG) shows continuous muscle activity at rest. Unlike other myotonic diseases, electrical discharges are high voltage and not lost by sleep and general anesthesia [3-4]. Almost all patients have action myotonia and percussion myotonia. Muscle stiffness is also seen during rest and sleep. SJS is a special clinical condition in which myotonia and continuous muscle activity coexist. Furthermore, continuous muscle activity can not be suppressed by general anesthesia and curare. However, sodium channel blocking agents are effective in suppressing muscle activity [1-4]. Another important feature of the disease is that it can develop malignant hyperthermia with general anesthesia [1-4]. In particular, patients should be encouraged to take the view of the neurologist in patients who are warned and planned to undergo surgery. Sometimes, mild CPK elevations are detected in laboratory tests. Apart from that, there are no special findings for the mentioned disease. Biochemical, histopathological, and cytogenetic studies have no significant benefit in diagnosis. Histopathologic studies have reported that neurogenic and myogenic findings as well as normal occurrences [3,4]. The most common diseases in which the diagnosis complex is caused by SJS are congenital myotonic dystrophy, myotonia congenita, and paramyotonia congenita [2-4]. However, special facial appearance, permanent muscle activity on ENMG, and joint contractures differ from other diseases [2-4]. Freeman-Sheldon Syndrome (FSS) and Marden-Walker Syndrome (MWS) should also be kept in mind in the differential diagnosis of SJS. In the FSS, there is a shrunken mouth that gives a whistle-looking appearance, as well as a smooth face, long philtrum, flexion in the

fingers of hands and foots, and ulnar deviation [7]. MWS is characterized with mental retardation, congenital joint deformities, and growth retardation [7]. The patients benefit from treatment with mexiletine and procainamide [3,4]. There are studies reporting that the use of carbamazepine is also beneficial [8]. Physiotherapy and occupational therapy, as well as medication, are also crucial. Botulinum toxin injection is another option to reduce muscle spasms. Surgical interventions can be performed for joint deformities in appropriate cases.

In conclusion, our case was diagnosed as SJS with progressive joint and bone deformities, special facial appearance, myotonic discharges on EMG, and continuous muscle activity which were presented to draw attention to the rare disease.

Scientific Responsibility Statement

The authors declare that they are responsible for the article's scientific content including study design, data collection, analysis and interpretation, writing, some of the main line, or all of the preparation and scientific review of the contents and approval of the final version of the article.

Animal and human rights statement

All procedures performed in this study were in accordance with the ethical standards of the institutional and/or national research committee and with the 1964 Helsinki declaration and its later amendments or comparable ethical standards. No animal or human studies were carried out by the authors for this article.

Conflict of interest

None of the authors received any type of financial support that could be considered potential conflict of interest regarding the manuscript or its submission.

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